Pulmonary Alveolar Microlithiasis

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A 28-year-old Iranian man who had lived in the United Kingdom for 3 years came to the outpatient clinic with a 3-year history of frequent cough, which occasionally produced a little sputum. He had never smoked and did not describe any other respiratory symptoms. Examination revealed sparse fine crepitations over all lung zones. Spirometry revealed a forced expiratory volume in 1 second of 2.85 L (73%) and a forced vital capacity of 4.01 L (86%). A roentgenogram of the chest revealed diffuse micronodular opacities resembling milky tuberculosis (Fig 1A). His tuberculin test gave a Heaf grade I reaction, and his sputum samples were negative for tuberculosis. The results of laboratory tests for blood count, biochemical profile, C-reactive protein, and erythrocyte sedimentation rate were all normal. An autoantibody screen produced negative results. Computed tomography of the thorax revealed extensive fine centrilobular nodularity throughout both lungs (Fig 1B), associated with ground-glass changes and septal thickening. There were small areas of subpleural calcification, particularly at both lung bases. He underwent bronchoscopy with transbronchial biopsy, which demonstrated numerous calcified bodies within the alveolar spaces (Fig 1C), many of which had a concentric laminated appearance. A pathognomonic finding of pulmonary alveolar microlithiasis was made.

Pulmonary alveolar microlithiasis is an autorecessive condition thought to involve mutations of the SLC34A2 gene. Fewer than 600 cases have been described worldwide. A chest x-ray film usually demonstrates fine micronodular opacities, and high-resolution computed tomography shows septal thickening, calcified micronodules, ground-glass changes, and subpleural calcification. Diagnosis is usually made as an incidental finding on chest roentgenography performed for unrelated reasons. Patients may remain asymptomatic with normal spirometric values or mild restriction. Symptoms can include exertional dyspnea, cough, and chest pain. The disease usually progresses to lung fibrosis, respiratory failure, and cor pulmonale although the pace of this progression varies.

Histology demonstrates laminated calcified bodies, with an onion skin–like appearance, usually ranging from 50 to 1,000 µm in diameter.

There is no effective therapy apart from lung transplantation.